G6PD gene

glucose-6-phosphate dehydrogenase

Normal Function

The *G6PD* gene provides instructions for making an enzyme called glucose-6-phosphate dehydrogenase. This enzyme, which is active in virtually all types of cells, is involved in the normal processing of carbohydrates. It plays a critical role in red blood cells, which carry oxygen from the lungs to tissues throughout the body. This enzyme helps protect red blood cells from damage and premature destruction.

Glucose-6-phosphate dehydrogenase is responsible for the first step in a chemical pathway that converts glucose (a type of sugar found in most carbohydrates) to ribose-5-phosphate. Ribose-5-phosphate is an important component of nucleotides, which are the building blocks of DNA and its chemical cousin RNA. This chemical reaction produces a molecule called NADPH, which plays a role in protecting cells from potentially harmful molecules called reactive oxygen species. These molecules are byproducts of normal cellular functions. Reactions involving NADPH produce compounds that prevent reactive oxygen species from building up to toxic levels within cells. The production of NADPH by glucose-6-phosphate dehydrogenase is essential in red blood cells, which are particularly susceptible to damage by reactive oxygen species because they lack other NADPH-producing enzymes.

Health Conditions Related to Genetic Changes

glucose-6-phosphate dehydrogenase deficiency

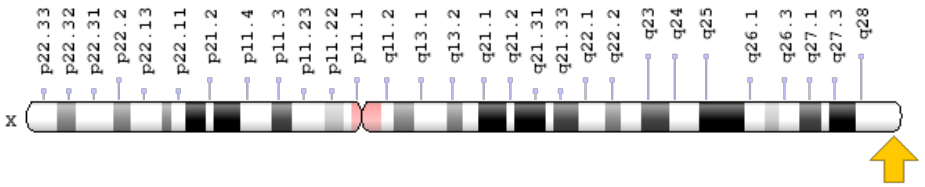
More than 140 mutations that cause glucose-6-phosphate dehydrogenase deficiency have been identified in the *G6PD* gene. Almost all of these mutations lead to changes in single building blocks (amino acids) that make up the glucose-6-phosphate dehydrogenase enzyme. These changes disrupt the normal structure and function of the enzyme or reduce the amount of the enzyme in cells.

Without enough functional glucose-6-phosphate dehydrogenase, red blood cells are unable to protect themselves from the damaging effects of reactive oxygen species. The damaged cells are likely to rupture and break down prematurely (undergo hemolysis). Factors such as infections, certain drugs, and ingesting fava beans can increase the levels of reactive oxygen species, causing red blood cells to undergo hemolysis faster than the body can replace them. This loss of red blood cells causes the signs and symptoms of hemolytic anemia, which is a characteristic feature of glucose-6-phosphate dehydrogenase deficiency.

Chromosomal Location

Cytogenetic Location: Xq28, which is the long (q) arm of the X chromosome at position 28

Molecular Location: base pairs 154,531,390 to 154,547,586 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- G6PD1
- G6PD_HUMAN

Additional Information & Resources

Educational Resources

- Chapter 20.5: Glucose 6-Phosphate Dehydrogenase Plays a Key Role in Protection Against Reactive Oxygen Species (Biochemistry, fifth edition, 2002) https://www.ncbi.nlm.nih.gov/books/NBK22389/

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28G6PD%5BTIAB%5D%29+OR+%28glucose-6-phosphate+dehydrogenase%5BTIAB%5D%29%29+AND+%28%28glucose-6-phosphate+dehydrogenase%5BMAJR%5D%29+OR+%28glucosephosphate+dehydrogenase%5BMAJR%5D%29+OR+%28d-glucose-6-phosphate:nadp++1-oxidoreductase%5BMAJR%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- GLUCOSE-6-PHOSPHATE DEHYDROGENASE http://omim.org/entry/305900

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_G6PD.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=G6PD%5Bgene%5D
- G6PD mutation database (University College London)
  http://www.bioinf.org.uk/g6pd/
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4057
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2539
- UniProt
  http://www.uniprot.org/uniprot/P11413

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